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Neonatal tuberous sclerosis

US, CT, and MR diagnosis of brain and cardiac lesions

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Abstract. Cortical, subcortical and subependymal tubers were more completely and more clearly depicted by MR than by CT and US in a 3-week-old infant. These lesions were best shown on T1-weighted images as areas of high signal intensity.

Case report

A 3-week-old girl was referred for evaluation of cardiac tumors diagnosed in utero at 36 weeks gestation and associated with arrhythmias necessitating Caesarean section. Postnatal 2D echocardiography showed two tumors. Diagnosis of tuberous sclerosis was suspected and the brain investigated. Brain sonography revealed several isoechogenic nodules along the floor of the lateral ventricules and adjacent to the foramen of Monro. CT scan demonstrated slight hyperdense (density: 40 UH), noncalcified periventricular nodules. Small ill-defined hyperdense areas (density: 40 UH) were observed in white matter and subcortical areas (Fig. 2).

MR imaging studies of the heart and the brain were performed to better determine the extent of the lesions (Siemens Magnetom operating at 1.5 Tesla). For the heart: ECG-gated MR imaging was obtained on T1-weighted spin-echo (SE) pulse sequences [echo time (TE): 20 msec and repetition time (TR): determined by heart rate]. MR easily depicted two cardiac tumors with intensity slightly higher or similar to that of the myocardium (Fig. 1).

In the brain, subependymal tubers appeared as bright nodules on T1-weighted SE images (TR: 600 msec, TE: 26 msec) and so called "proton-density" images (TR: 2600 msec. TE: 26 msec). Cortical, subcortical and white matter lesions were otherwise detected as patches of increased signal intensity (Fig. 3). On T2-weighted images (TR: 2600 msec, TE: 140 msec); all those lesions appeared slightly hypointense and were much more difficult to detect. Other investigations (electroencephalogram, renal sonography, skull radiograph, Wood's light exploration of the skin, ophthalmic examination ...) were normal.

Discussion

In TS the cellular differentiation is disturbed to a varying degree [1] and, classically, there is a triad of mental retardation, seizures and adenoma sebaceum or depigmented areas. However, manifestations of the disease have a wide spectrum and many incomplete forms exist. Therefore the incidence of TS is difficult to evaluate and is probably higher than 1 in 10,000 to 350,000 [2].

Tuberous sclerosis follows an autosomal dominant heredity with variable expression. 50% to 80% of the childhood cases are considered as spontaneous mutations [2, 3, 4].

Intracranial manifestations of TS include subependymal, cortical and subcortical white matter tubers, consisting of a very cellular dysplastic mass, composed mainly of abnormal giant astrocytes [2, 3].

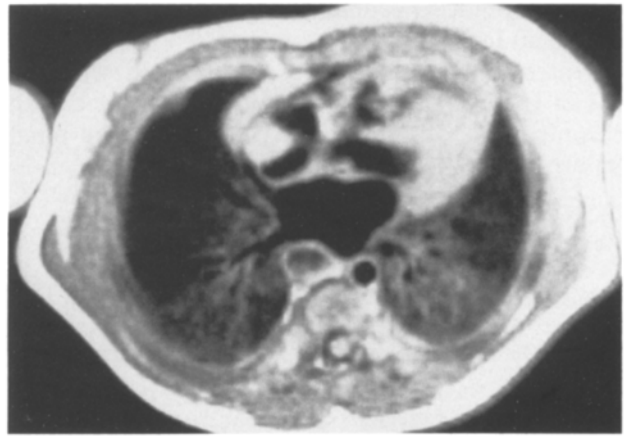

Fig. 1. Cardiac axial SE MR scan: slightly hyperintense mass in the right atrium

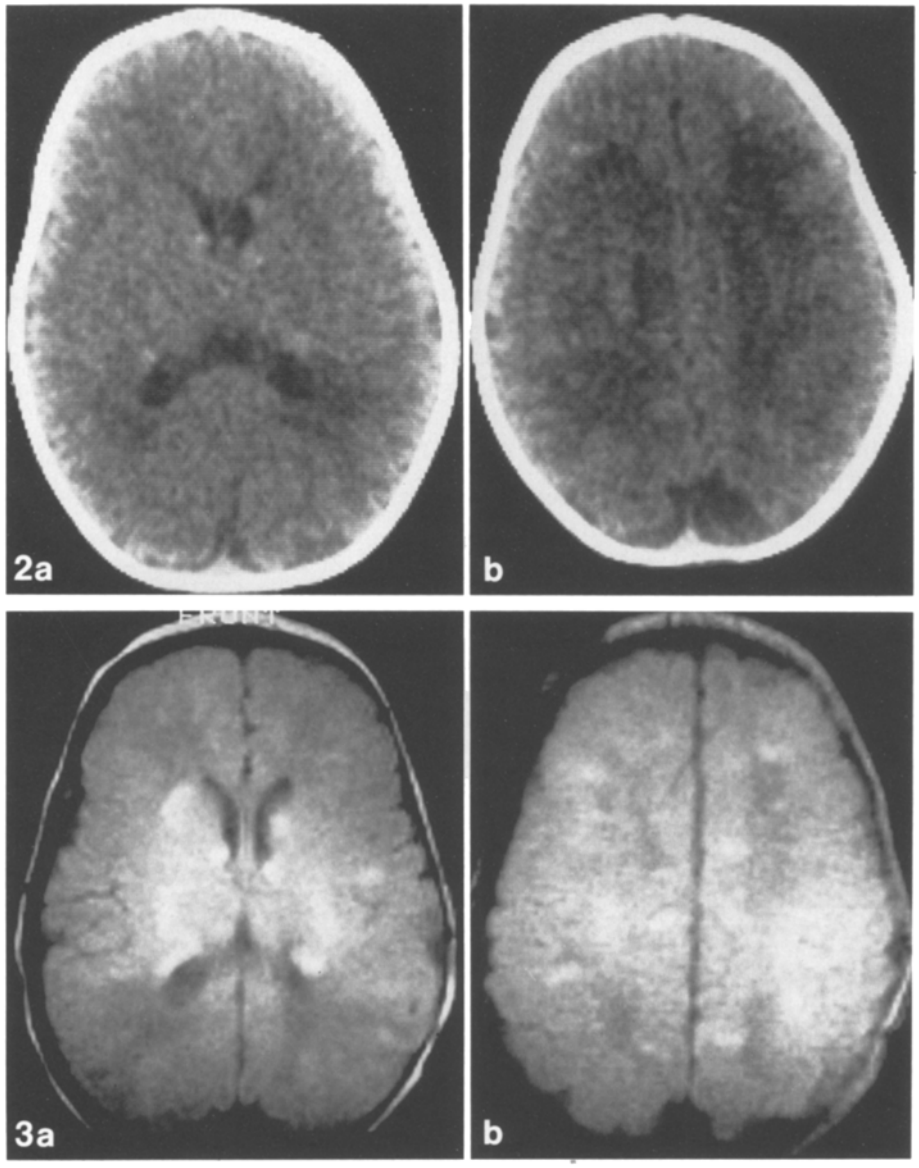

Fig. 2a, b. CT scan without contrast material. **a** small periventricular subependymal tubers of high density, **b** slightly hyperdense areas within subcortical white matter presumably representing intraparenchymal tubers

Fig. 3a, b. Axial MR scan (SE 2600/26). **a** hyperintense subependymal and subcortical tubers. Diffuse hyperintense lesion in the white matter suggestive of heterotopic areas. Beginning of myelination in the posterior limb of internal capsule, as hyperintense spots, **b** punctiform hyperintense lesion disseminated in the cortical-subcortical areas consistent with small tubers or heterotopic clusters of giant cells

Heterotopias of neuronal or glial cells, myelination defects, gliosis and spongiosis are other lesions observed in TS. Hydrocephalus due to obstruction of CSF flow may be caused by tubers or their malignant transformation. The occurrence and intensity of iron-calcium deposits in the lesions are age dependent.

Ultrasound did not clearly differentiate noncalcified subependymal tubers from the normal surrounding underlying brain tissue, and failed to outline intraparenchymal tubers. By contrast, CT detected at least 10 noncalcified periventricular tubers. These appeared slightly hyperdense compared to the adjacent unmyelinated white matter.

Subcortical and white matter lesions, consisting of ill-defined high density areas represented probably non-calcified intraparenchymal tubers. In older patients, these are usually described as lesions of low density attributed to parenchymal tubers and the surrounding area of demyelination [3, 4]. MR demonstrated more lesions than US and CT and with better definition. On T1-weighted and "proton density" SE sequences all the lesions had quite the same MR signal characteristics and appeared as hyperintense areas (whereas they are relatively isosignal in the adults [4, 5]). The so called "proton density" SE sequences used in the adults correspond in fact to T1-weighted images in the newborn brain, owing to the long T1 relaxation time.

Subependymal hyperintense nodules are consistent with paraventricular tubers. Hyperintense patches and punctiform foci disseminated in corti-